



Overview

Points To Remember About Epidermolysis Bullosa

- Epidermolysis bullosa is a group of diseases in which the skin is easily injured, causing painful blisters to form. These blisters can cause serious problems if they become infected.
- Some people with the condition have a mild form with few blisters. Others may have many blisters.
- A doctor can identify the disease by taking a small piece of skin and looking at it under a microscope.
- Treatment includes proper skin care to prevent blisters, treating blisters and infections, and a good diet. Surgery may be needed in more severe cases.
- Talk to your doctor about how to treat symptoms when they occur.

Epidermolysis bullosa is a group of diseases in which even minor rubbing of the skin can cause blistering. In severe cases, blisters may also develop inside the body, such as in the mouth, esophagus, stomach, intestines, upper airway, bladder, and genitals.

If you have epidermolysis bullosa, you received faulty genes from either one or both parents. You can have the disease even if your parents do not show symptoms of it. A genetic counselor can test for disease genes, provide information on the likelihood of passing the defective gene to your children, and provide advice on future childbearing.

Who Gets

Epidermolysis bullosa occurs in all racial and ethnic groups and affects males and females equally. The disease is not always evident at birth. Milder cases of epidermolysis bullosa may appear when a child crawls, walks, or runs, or when a young adult is very active.

Types

The different forms of epidermolysis bullosa include:

- **Epidermolysis bullosa simplex**—tissue separation and blistering occur in the top layer of the skin (epidermis).
- **Junctional epidermolysis bullosa**—tissue separation and blistering occur in the top portion of the basement membrane (area between the skin layers), due to problems in attachment between the epidermis and basement membrane.
- **Dystrophic epidermolysis bullosa**—tissue separation and blistering occur in the basement membrane due to problems in attachment between the epidermis and dermis (the inner skin layer).
- **Epidermolysis bullosa acquisita**—a rare autoimmune disorder where the body attacks its own anchoring structures in the skin.

Symptoms

Epidermolysis bullosa is indicated by skin blisters, although each disease type can have somewhat different symptoms:

- **Epidermolysis bullosa simplex** usually begins with blistering at birth or shortly afterward. In a mild subtype called Weber-Cockayne, blisters are usually found on the feet and hands. In other subtypes, the blisters occur over the entire body. Other signs may include thickened skin on the palms of the hands and soles of the feet; rough, thickened, or absent fingernails or toenails; and blistering inside the mouth. Less common signs include slow growth; blisters in the esophagus; anemia; scarring of the skin; and small, white skin bumps.
- **Junctional epidermolysis bullosa** is usually severe. The most serious form can be life-threatening because large, open blisters on the face, trunk, and legs may become infected or produce severe dehydration due to fluid loss. Survival is also threatened by blisters in the esophagus, upper airway, stomach, intestines, urinary system, and genitals. Other signs found in both severe and milder forms are rough and thickened or absent fingernails and toenails; a thin appearance to the skin; blisters on the scalp or loss of hair with scarring; malnutrition; anemia; slow growth; blisters inside the mouth and nose; and poorly formed tooth enamel.
- **Dystrophic epidermolysis bullosa** has slightly different symptoms, depending upon whether the disease gene is inherited from one or both parents.
 - **Dominant subtype** is where the disease gene is passed down from only one parent.

In some cases, blisters may appear only on the hands, feet, elbows, and knees; nails usually are shaped differently; small, white bumps may appear on the skin of the trunk and limbs; and there may be involvement of the soft tissues, especially the esophagus.

- **Recessive subtype** is where the disease gene is passed down from both parents. This subtype is characterized by blisters over large areas of the body; loss of nails or rough or thick nails; scars that leave a pit in the skin; small, white bumps on the skin; itching, anemia; and slow body growth.
 - In milder cases of this subtype, blisters may appear only on the hands, feet, elbows, and knees; nails usually are shaped differently; small, white bumps may appear on the skin of the trunk and limbs; and there may be involvement of the soft tissues, especially the esophagus.
 - Severe forms of this subtype may lead to eye damage; tooth loss; blistering inside the mouth and gastrointestinal tract; and fusing together of the fingers or toes. There is also a high risk of developing skin cancer on the hands and feet. This cancer tends to grow and spread faster in people with epidermolysis bullosa than in those without the disease.

Causes

If you have epidermolysis bullosa, you received faulty genes from either one or both parents. You can have the disease even if your parents do not show symptoms of it.

Specific causes depend on the form of the disease:

- In **epidermolysis bullosa simplex**, there is usually a defect in genes inherited from one parent. The faulty genes are those responsible for formation of an anchoring protein in the top layer of skin. As a result, the skin splits in the epidermis, producing a blister.
- In **junctional epidermolysis bullosa**, there is a defect in the genes inherited from both parents. These genes normally promote the formation of structures that anchor the top layer of the skin (epidermis) to the basement membrane (area between the epidermis and dermis, the bottom-most skin layer). The defect leads to tissue separation and blistering in the upper part of the basement membrane.
- In **dystrophic epidermolysis bullosa**, there are defective genes inherited from either one or both parents. The result is that the anchoring structures between the epidermis and the dermis are either absent or do not function.
- **Epidermolysis bullosa acquisita** sometimes occurs following drug therapy for another condition. In most cases, the cause is unknown.

Diagnosis

Dermatologists can identify where the skin is separating to form blisters. To determine the type of epidermolysis bullosa, a small sample of skin is examined under a microscope to see if

certain proteins are missing or reduced, or if there are problems in skin structure.

Defective genes can also be identified in epidermolysis bullosa patients and their family members. Prenatal diagnosis can now be accomplished as early as the 10th week of pregnancy.

Treatment

There is no cure for epidermolysis bullosa, although there are medicines to help prevent infection and to reduce discomfort. Consult your health care professional about the best options for you.

Goals of treatment include [preventing blisters, caring for blistered skin, treating infection, and treating nutritional problems](#).

Surgery may be an option in some cases:

- Severe forms of dystrophic epidermolysis bullosa may cause the esophagus to narrow, requiring it to be widened so that food can travel from the mouth to the stomach.
- People who are not getting proper nutrition may need a tube that places food directly in the stomach.
- People whose fingers or toes are fused together may require surgery to release them.

Who Treats

You may see one of the following specialists:

- Dermatologists can diagnose epidermolysis bullosa by identifying where the skin is separating to form blisters. To determine the type of epidermolysis bullosa, a small sample of skin is examined under a microscope to see if certain proteins are missing or reduced, or if there are problems in skin structure.
- Genetic counselors can test for disease genes, provide information on the likelihood of passing the defective gene to your children, and provide advice on future childbearing.
- Dietitians can find recipes for food that is nutritious and easy to consume, and recommend diets to prevent constipation, diarrhea, or painful elimination.

Living With

You should try to keep blisters from forming and prevent infection when blisters occur. If you have moderate and severe forms of epidermolysis bullosa, you not only need to care for and protect your skin, but you may also have many complications that require psychological support. Doctors, nurses, social workers, clergy members, psychologists, dietitians, and patient and

parent support groups can assist with care and provide information and emotional support.

- Preventing blisters. In many forms of epidermolysis bullosa, blisters will form with the slightest pressure or friction. You should not let your disease prevent you from cuddling your baby, who needs to feel a gentle human touch and affection. There are a number of ways you can protect the skin from injury:
 - Avoiding overheating by keeping rooms at an even temperature.
 - Applying lotions to the skin to reduce friction and keep the skin moist.
 - Using simple, soft clothing that you can easily get on or off your child.
 - Using sheepskin on car seats and other hard surfaces.
 - Wearing mittens at bedtime to help prevent scratching.
- Caring for blistered skin. When blisters appear, you will need to reduce pain or discomfort, prevent loss of body fluid, promote healing, and prevent infection.
 - The doctor may prescribe a mild pain reliever during bandage changes. Bandages that are sticking to the skin may be removed by soaking them in warm water. You can use mild soaps during daily bathing, although you should clean small areas at a time.
 - Blisters can become quite large and create a large wound when they break. A medical professional can tell you how to safely break a new blister to keep the top skin in place. After opening and draining the blister, the doctor may suggest applying an antibiotic to the area before covering it with sterile, nonsticking gauze pad that is held with gauze strips (not tape). The doctor may recommend keeping the broken blister uncovered in milder cases or if it's in an area that is difficult to cover.
 - A somewhat moist environment helps with healing. However, a covering may be needed to absorb heavy fluid from blister areas, which may otherwise irritate your skin. Your doctor can recommend the best coverings to use for your specific wound.
- Treating infection. The chances of skin infection can be reduced by good nutrition and by careful skin care with clean hands and sterile materials. Your doctor may recommend antibiotic creams and soaks to further reduce the risk.
 - However, you can still develop infection even if you do all of these things. Signs of infection are redness and heat around an open area of skin, pus, excessive crusting on the wound surface, a red line or streak under the skin that spreads away from the blistered area, a wound that does not heal, and/or fever or chills. The doctor may prescribe a specific soaking solution or an antibiotic cream or pill. Wounds that are not healing may be treated by special coverings.
- Treating nutritional problems. Some people with epidermolysis bullosa may have blisters in the mouth and esophagus, which may make it difficult to chew and swallow food and drinks. Dietitians can find recipes for food that is nutritious and easy to consume, and recommend diets to prevent constipation, diarrhea, or painful elimination.
 - If breast or bottle feeding produces blisters, infants may be fed using a preemie nipple (a soft nipple with large holes), a cleft palate nipple, an eyedropper, or a syringe. When the baby is old enough to take in food, adding extra liquid to finely mashed food makes it easier to swallow. Soups, milk drinks, mashed potatoes, custards, and puddings can be given to young children. However, food should never be served too hot.

Research Progress

Current research focuses on finding faulty genes and their effect on the tissues, copying genes, correcting faulty genes or replacing them with healthy ones, and screening for disease genes. Researchers are also looking at healthy skin for clues as to why development goes wrong to cause diseases.

Related Resources

U.S. Food and Drug Administration

Toll free: 888-INFO-FDA (888-463-6332)

Website: <https://www.fda.gov>

Drugs@FDA at <https://www.accessdata.fda.gov/scripts/cder/daf>. Drugs@FDA is a searchable catalog of FDA-approved drug products.

Centers for Disease Control and Prevention, National Center for Health Statistics

Website: <https://www.cdc.gov/nchs>

The Dystrophic Epidermolysis Bullosa Research Association of America, Inc.

Website: <http://www.debra.org>

National Society of Genetic Counselors

Website: <http://www.nsgc.org>

American Academy of Dermatology

Website: <https://www.aad.org>

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at NIAMSInfo@mail.nih.gov.

- [Asian Language Health Information](#)
- [Spanish Language Health Information](#)

Join a Clinical Trial

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Related Information

[Questions and Answers about Epidermolysis Bullosa](#)

[Healthy Skin Matters: Kids' Fact Sheet](#)

View/Download/Order Publications

[Epidermolysis Bullosa, Easy-to-Read Fast Facts](#)